

CONGENITAL MITRAL STENOSIS

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Only 14 examples of congenital mitral stenosis uncomplicated by other cardiovascular abnormality have been reported (Summons, 1906; Newns, 1938; Emery and Illingworth, 1951; Maxwell and Young, 1954; Braudo *et al.*, 1957; Bower *et al.*, 1953; Starkey, 1959). In the majority of accounts of this subject, mitral stenosis has been complicated by other cardiac anomalies. Thus, of the 9 patients described by Ferencz *et al.* (1954), 8 had significant associated cardiac lesions, and of 34 patients quoted by them from other sources, only 8 had an uncomplicated lesion.

The descriptions of the clinical features of previously reported cases are confusing because the majority had additional cardiac lesions. We are therefore describing four patients with isolated congenital mitral stenosis to clarify the clinical features, to stress the poor prognosis, and to discuss surgical treatment.

CASE REPORTS

Case 1. This boy was well until the age of 2½ years when he developed breathlessness on effort, cough, and chest and limb pains. His parents were second cousins, his father had had rheumatic fever, but there was no other family history of disease of the cardiovascular or other systems. There were three normal siblings. Pregnancy and delivery were normal and his birth weight was 6 lb. (2·7 kg.). He was first seen at the Hospital for Sick Children, Great Ormond Street, in February 1952, at the age of 3 years 8 months. His weight was 33½ lb. (15·2 kg.) (45th percentile) and height 37½ in. (94·75 cm.) (10th percentile). There was no cyanosis or congestive failure; the radial and femoral pulses were normal, and the systolic blood pressure was 125 mm. Hg. There were no thrills, and the apex beat was in the nipple line. On auscultation the first sound at the apex and the second sound in the left second space were accentuated; there was no opening snap; and a systolic murmur was present at the apex. The cardiogram (Fig. 1) showed normal axis and P waves 4 mm. tall and peaked in lead I, and inverted in leads V1 and V3. Radiographs (Fig. 2) indicated right ventricular and left atrial enlargement and increased pulmonary vascular markings. A provisional diagnosis of mitral stenosis was made. He was treated with digoxin and his symptoms responded well.

He was seen again in December 1953, at which time a diastolic murmur with presystolic accentuation had appeared; in addition there was a diastolic murmur along the left sternal border which was thought to be due to pulmonary regurgitation.

Right heart catheterization was performed at the National Heart Hospital in April 1954, and the findings are summarized in the Table. There was a moderate rise of the pulmonary arterial pressure (mean 50 mm. Hg). As the mean pulmonary wedge pressure was raised to 23 mm. Hg, left atrial puncture was planned but was abandoned because of severe bronchospasm during induction of general anaesthesia.

By December 1959, at the age of 11½ years, his exercise tolerance had deteriorated and was limited to a few yards of walking on the flat. Crepitations were present at both lung bases, the liver was two and a half fingers below the costal margin, and a diastolic thrill was felt at the apex. The blood pressure was 125/65 mm. Hg. A chest radiograph showed moderate cardiac enlargement and marked enlargement of the left atrium and proximal pulmonary vessels; the aorta was small; and the lung fields showed coarse reticular type of shadowing, Kerley's lines, and hæmosiderosis.

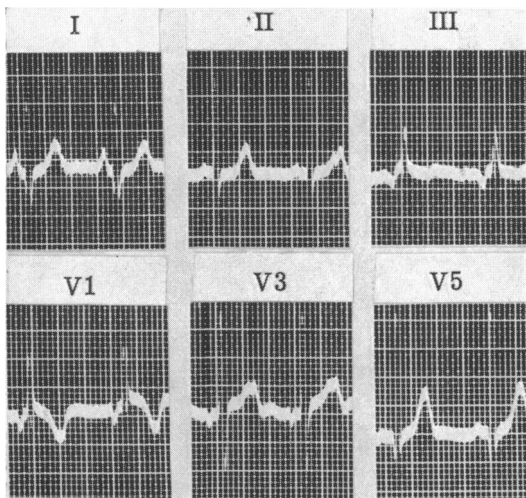


FIG. 1.—Case 1: aged $3\frac{1}{2}$ years. Tall, peaked P waves in lead I.

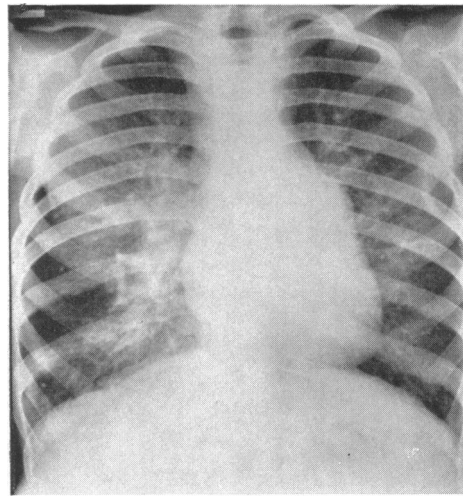


FIG. 2.—Case 2. Right ventricular and left atrial enlargement and increased pulmonary vascular markings.

Mitral valvotomy was performed at University College Hospital in January 1960. The lung was found to be adherent to the pericardium which was thickened. There was a large pericardial defect just behind the phrenic nerve. The pulmonary artery was enlarged and tense. The left atrial appendage was large and bulged laterally through the pericardial defect. The mitral valve orifice would not admit a finger, but when the finger was thrust through the opening the valve stretched without splitting. The aortic cusp of the mitral valve was supple. Through an opening in the tip of the left ventricle, a Tubb's dilator was passed through the valve and opened in three stages; the first two produced no regurgitation but the third dilatation produced a considerable regurgitant jet. After the third stage the dilator was judged to be open a little less than 4 cm.

Eleven days after the valvotomy a chest radiograph showed a further increase in the cardiac enlargement and in the pulmonary vascular markings. A big improvement was noted after eight weeks, and the patient could walk about one and a half miles. Eleven weeks after valvotomy he was able to play football, and at his last visit 17 months after valvotomy the improvement had been maintained, but signs of mitral regurgitation were present.

TABLE
CATHETERIZATION DATA

Cases	Age (years)	Pressures mm. Hg						Cardiac output (l./min./m. ²)	Pulmonary vascular resistance units/m ²
		RA mean	Mean pulmonary wedge	RV	PA	PA mean	FA		
1	6	0	23	62/0	67/39	50	95/55	5.3	5
2	4	9	23	75/4	75/42	57	—	4.6*	7
3	11/12	6	—	132-166 1	141-159 58	85	86/50	3.0†	18‡
4	11/12	9	16	90/5	90/50	60	85/53	3.1†	14

* Assumed arterial saturation 95 per cent. Assumed resting oxygen consumption 180 ml./min./m.²

† Assumed resting oxygen consumption 200 ml./min./m.²

‡ Assumed left atrial mean pressure 30 mm. Hg.

Case 2. This boy was first seen at Guy's Hospital, in September 1958, at the age of 3 years and 4 months, with a history of a recurrent cough, worse in the winter for two and a half years, and breathlessness on exertion. A provisional diagnosis of ventricular septal defect had been made. He was born at 36 weeks gestation weighing 5 lb. (2.26 kg.). On examination his weight was 28 lb. (12.7 kg.) (under 3rd percentile). There was a marked thrust over the right ventricle and a diastolic shock over the pulmonary area with a loud second sound. At the apex, a diastolic thrill and rumble were present. There was no opening snap. The cardiogram (Fig. 3) showed normal P waves and right ventricular hypertrophy, with R in V1 of 40 mm. and S

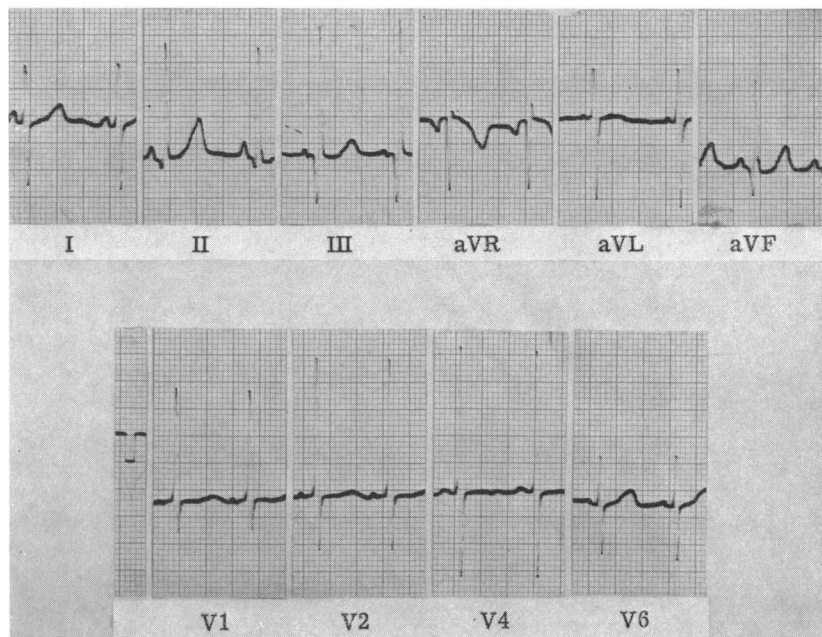


FIG. 3.—Case 2: aged 3½ years. Right ventricular hypertrophy.

in V6 of 20 mm. Radiographs indicated right ventricular hypertrophy and a dilated pulmonary artery, and the lung markings suggested venous congestion. Barium swallow did not show enlargement of the left atrium, though a radiograph one and a half years previously (Fig. 4), was suggestive of mitral disease. Right heart catheterization was performed from the right saphenous vein under inhalation anaesthesia. The results are summarized in the Table. No shunt was demonstrated. The high mean wedge pressure of 23 mm. Hg and moderately severe right ventricular and pulmonary hypertension confirmed the clinical findings of mitral stenosis. An angiocardiogram was done when the catheter was in the right pulmonary artery, but no enlargement of the left atrium was demonstrated.

A closed valvotomy was performed. At operation, the right ventricle was large and the pulmonary artery was large and tense. The aorta was a little narrowed in the region of the ductus arteriosus. However, this was judged to be functionally unimportant since there was good pulsation in the descending aorta and anastomotic vessels were not present. The left atrial appendage was considerably enlarged. The left lung showed moderate pigment changes. A powerful filling thrill could be felt over the left ventricle. The mean pressure in the left atrium was 39 cm. of saline. When the appendage was opened it contained no clot but the lining was thick and opaque. Digital exploration revealed no regurgitation. The valve orifice was about 0.5 cm. × 0.4 cm. and was firm. It resisted finger pressure and retrograde dilatation was performed. The valve split freely and widely and apparently along normally placed commissures and gave a large full-sized opening without regurgitation. The heart was not distressed after this, and the mean pressure in the left atrium was 41 cm. of saline. By the time the chest was being closed the heart action was once more good.

Histological examination of the left atrial appendage showed fibro-elastic thickening of the endocardium. The post-operative course was one of recurrent right heart failure, and it was clear from the physical signs

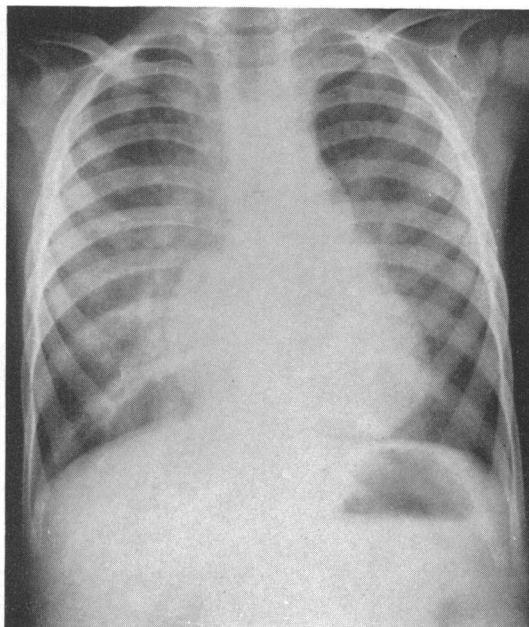


FIG. 4.—Case 2. Right ventricular hypertrophy, dilated pulmonary artery, and pulmonary venous congestion.

that mitral regurgitation was present. The heart size radiographically increased from 9 cm. to 11 cm. following operation.

A year after the operation he was attending school but had had four attacks of bronchitis. Mitral systolic and diastolic murmurs were present and full length. The electrocardiogram had not changed from before operation. One year later, although he had some breathlessness on exertion, there had been an increase in exercise tolerance but no change in his clinical or radiological signs.

Case 3. This infant was admitted to the Royal Aberdeen Hospital for Sick Children in February 1960, when he was 7 months old, with a history of cough, refusal to feed, and loss of weight for 14 days. He was a third child, full term, weighing 8 lb. 4 oz. (3.7 kg.) at birth and 15 lb. 10 oz. (7.1 kg.) (10th percentile) on admission. He was not cyanosed or in heart failure, but a loud systolic murmur maximal at the apex was heard all over the præcordium, in the back, and in the left axilla. The chest radiograph showed a normal-sized heart but the pulmonary vascular markings were increased. An upper respiratory tract infection was diagnosed, and he was treated with penicillin by intramuscular injection for eight days. Six weeks later he was re-admitted with a further respiratory infection which again responded to penicillin. At the age of 10 months he was admitted for a third time in heart failure. Blood cultures grew *Staph. aureus* and *Strep. viridans*. His fever resolved with antibiotic therapy and the blood culture became sterile. His heart failure diminished on treatment with digitalin (0.1 mg) which was discontinued after three weeks because of vomiting. Since his first admission chest radiograph showed progressive cardiac enlargement. He was transferred to Guy's Hospital on June 15, 1960 with a provisional diagnosis of ventricular septal defect.

On admission he was pale, afebrile, and in heart failure. At the apex the first sound was loud, there was an opening snap and a rumbling diastolic murmur. The electrocardiogram (Fig. 5) showed tall peaked P waves in lead II and right ventricular hypertrophy. Radiologically (Fig. 6) the cardio-thoracic ratio was 8/14 with enlargement of the left atrium confirmed by fluoroscopy, and the lungs showed increased pulmonary vascular markings. His hæmoglobin was 6.1 g. per 100 ml. (39%), hæmatocrit 18 per cent, and blood culture sterile. A diagnosis was made of congenital mitral stenosis with heart failure. He was treated with digoxin, chlorothiazide, and blood transfusion. His condition improved, the hæmoglobin rising to 9.7 g. per 100 ml. (62%).

Right heart catheterization was done on June 27, 1960, under sedation. The results, summarized in the Table, showed right ventricular and pulmonary hypertension with pulsus alternans. No shunt was

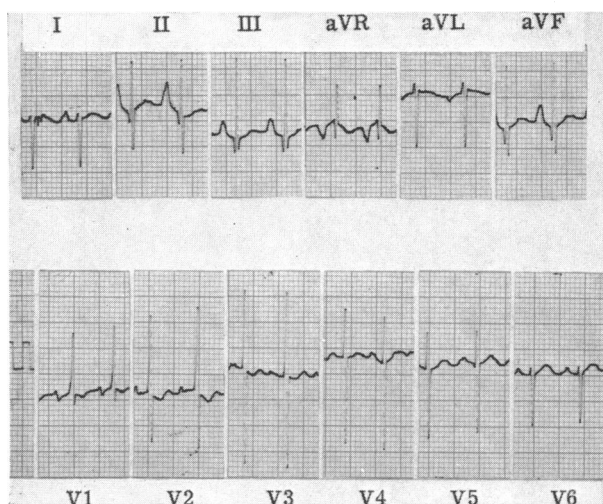


FIG. 5.—Case 3: aged 11 months. Tall peaked P waves in lead II and much right ventricular hypertrophy.

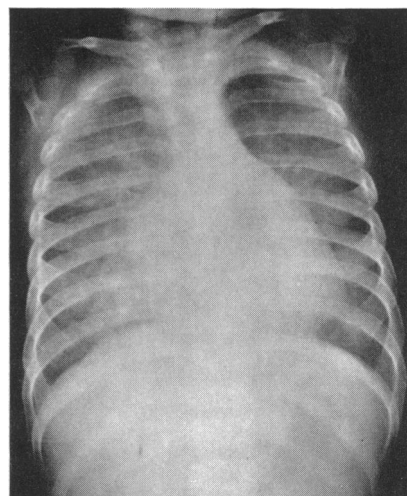


FIG. 6.—Case 3. Cardiomegaly, enlargement of the left atrium, and increased pulmonary vascular markings.

demonstrated either by oxygen saturations or by dye curves. On breathing oxygen, the femoral artery saturation rose from 92.5 per cent to 100 per cent and the pulmonary arterial pressure fell from 159/58 to 94/37 mm. Hg with a simultaneous femoral artery pressure of 94/45 mm. The pulsus alternans in the right ventricle was abolished and the arterio-venous oxygen difference fell from 6.7 to 4.7 vol. per cent. As no wedge pressure was obtained the pulmonary vascular resistance could not be calculated, but assuming a left atrial pressure of 30 mm. Hg the pulmonary vascular resistance would be about 18 units/m.²

On the clinical evidence and these findings, plans were made for mitral valvotomy under direct vision to be performed within the next few days. It was also decided that as oxygen had lowered the pulmonary artery pressure, he should be nursed in an oxygen tent before operation. However, two days later he developed sudden and severe pulmonary oedema which was unrelieved by medical treatment. An emergency closed valvotomy was therefore performed.

Left thoracotomy was performed through the 5th rib bed. The lung was brick red and turgid, the right heart was grossly dilated, the pulmonary artery and left atrium were both large and tense, and the left ventricle was small. A little finger in the left atrium failed to identify the mitral orifice. A sound passed from the left ventricle entered a mitral orifice about 4 mm. in diameter against the lateral side of the left ventricular cavity. A retrograde dilator was passed through this hole rupturing the membrane. Initially heart action was good but after three minutes it ceased, and resuscitation was unavailing. Necropsy showed partial relief of severe mitral stenosis without production of gross regurgitation. The left atrium was dilated with marked thickening of the wall, and the right ventricle also showed dilatation and hypertrophy. The pulmonary vessels were dilated and the lungs firmer than normal. No other congenital lesion was demonstrated. The pleurae and air passages were normal.

The sudden failure two days after oxygen therapy seems more than coincidental. The drop in pulmonary arterial pressure noted at catheterization, though relieving the right heart, may have increased the pulmonary flow and precipitated pulmonary oedema.

Case 4. This infant was admitted to the Royal Aberdeen Hospital for Sick Children at 4 months of age because of a respiratory infection and was found to have a loud systolic murmur. He was of special interest since his mother was the sister of Case 3. He was born at 37 weeks weighing 5 lb. 4 oz. (2.37 kg.). At 10 months he had developed a diastolic murmur and had failed to thrive (weight 13 lb. (5.89 kg.), less than the 3rd percentile). A chest radiograph indicated enlargement of the left atrium, and the electrocardiogram showed P waves 4 mm. tall and the pattern of marked right ventricular hypertrophy. A diagnosis of congenital mitral stenosis was made, and he was transferred to Guy's Hospital. Right heart catheterization (see Table) revealed a raised pulmonary arterial pressure (mean 60 mm.) and a raised pulmonary wedge pressure (mean 16 mm. Hg).

At open operation under hypothermic by-pass the valve orifice was found to be 4 mm. in diameter. This split fully through the lateral commissure but was resistant medially. On inspecting the under side of the valve at this point the papillary muscles were seen to run directly into the valve commissure as a solid band without intervening chordæ tendinæ. The commissure was then opened by sharp dissection, taking care to ensure adequate muscular support for each valve cusp.

Gross pulmonary hypertension persisted after valvotomy. Although good heart action could be maintained with a flow rate down to 100 ml. a minute, heart action failed when by-pass was discontinued.

At necropsy, apart from mitral stenosis, the only other abnormality was fibro-elastosis of the left atrium. Histologically the pulmonary arteries and arterioles were reported to be normal (Dr. R. Spector).

DISCUSSION

The diagnosis of congenital mitral stenosis does not exclude the possibility of endocardial fibro-elastosis, and in three of the patients this was demonstrated in the left atrial biopsy or at necropsy. We have no doubt, however, that in these patients it was the valve lesion, and not endocardial or myocardial pathological changes that determined the severity of the disease.

Isolated congenital mitral stenosis is not common. The patients now described are the only four with this lesion found in the records of the last 10 years of a general hospital (Guy's) and a Children's Hospital (Great Ormond Street), both having a special interest in congenital heart disease. Mitral stenosis, however, is less of a rarity when associated with other left-sided congenital lesions (Craig, 1949; Lev, 1952; Noonan and Nadas, 1958). An incidence of 4.3 per cent was found by Ferencz *et al.* (1954), in a series of 210 consecutive necropsies in children with congenital heart disease, but as only one of their nine patients had the isolated lesion, the corrected incidence was 0.48 per cent. The Hospital for Sick Children, Toronto, has only five cases with this diagnosis, of which three with full investigations have other congenital lesions, while the other two have clinical evidence of isolated mitral stenosis (J. D. Keith, 1961, personal communication). Our object is not to stress the rarity of the condition but to emphasize that the diagnosis is not difficult if the possibility is entertained.

All four of our patients had characteristic diastolic murmurs and one had an opening snap. This is not unexpected for it is difficult to imagine that a severe obstruction at the mitral valve can occur without a murmur at some time. No characteristic murmur was noted in 6 of the 14 patients with isolated mitral stenosis previously reported. These children, however, may present in failure so that a murmur may not be heard until the failure is treated. Ferencz *et al.* (1954) stressed the rarity of diastolic murmurs but this may be because the mitral stenosis was associated with other major congenital defects in most of their patients. We have, however, heard the characteristic murmur when mitral stenosis is not the only lesion. In the light of our own experience and that of others (Summons, 1906; Newns, 1938; Emery and Illingworth, 1951; Bower *et al.*, 1953; Kjellberg *et al.*, 1959; Hauck *et al.*, 1959; Starkey, 1959), we hesitate to diagnose congenital mitral stenosis without a diastolic murmur. On one occasion we made an erroneous diagnosis of mitral stenosis in the absence of a murmur in a child in heart failure with a large left atrium and severe pulmonary venous congestion confirmed by catheterization. Mitral valvotomy was planned, but at operation fibro-elastosis of the left ventricle was found without mitral stenosis, and this was later confirmed at necropsy.

Although previous reports contain scanty data on the cardiograms, it was helpful in our four patients, three showing abnormal P waves and three excessive right ventricular preponderance. Radiology showed pulmonary venous congestion in three of our patients. The increased vascular markings may be mistaken for the pleonæmia of a left-to-right shunt, and this was so in two of our patients who were originally diagnosed as ventricular septal defects. The left atrium was seen to be enlarged in three patients. The film of the second patient (Fig. 4) suggested enlargement of the left atrium but this was not confirmed by fluoroscopy or angiocardiography though the chamber was found to be considerably enlarged at operation. Angiocardiography was not done in the three who showed unequivocal left atrial enlargement by radiography. Right heart catheterization showed

high "wedge" pressures in the three patients in whom the pulmonary artery was entered. Thus, the findings of a mitral diastolic murmur, electrocardiographic evidence of right ventricular hypertrophy with abnormal P waves, and the radiological appearances of pulmonary venous congestion and left atrial enlargement are the important points in making a provisional diagnosis. Cardiac catheterization is most helpful in confirming the diagnosis.

SURGICAL MANAGEMENT

The results of mitral valvotomy in acquired mitral stenosis are good. In the congenital variety, however, there are additional problems. A conventional digital palpation and assessment of the valve mechanism may be difficult or impossible because of the small size of the heart and left atrial appendage. Also some of these valves are malformed and present as a diaphragm with a hole (Starkey, 1959). Moreover, associated fibro-elastosis may make them tough and elastic.

The combination of a small atrial appendage and the ill-defined tough orifice means that the use of a retrograde dilator is likely to be necessary to achieve a valve split. In rheumatic mitral stenosis, this instrument has the effect of splitting the valve along its commissures, but in congenital mitral stenosis it is likely to inflict crippling or fatal mitral regurgitation. A special child-size expanding dilator may be a useful and a safer instrument, and one of these is at present under construction. However, it is clear from these patients under review and from the experience of other surgeons (Starkey, 1959), that an open-heart technique offers the best chance of identifying the valve anatomy. In this way it should be possible to relieve the obstruction and at the same time identify and preserve the delicate chordæ tendinæ which ensure competence of the valve.

Because of the poor prognosis of untreated patients we wish to stress the need for prompt diagnosis, since the available evidence suggests that the outlook is improved by surgical treatment.

SUMMARY

The clinical, radiological, and electrocardiographic findings of four patients with isolated congenital mitral stenosis are described and discussed.

Isolated congenital mitral stenosis is rare; more commonly additional cardiac abnormalities are present, especially those involving the left side of the heart.

The differential diagnosis from ventricular septal defect is emphasized. In both conditions there may be radiological evidence of increased pulmonary vascular markings.

The poor prognosis can be improved by mitral valvotomy. Open-heart surgery is the method of choice in order to preserve mitral competence.

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